STATE OF WASHINGTON
WASHINGTON STATE BOARD OF HEALTH
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May 8, 2002

TO: Washington State Board of Health Members

FROM: Dr. Tom Locke, Board Member and Co-chair of the Newborn Screening Advisory Committee
Dr. Maxine Hayes, State Health Officer and Co-chair of the Newborn Screening Advisory Committee

RE: NEWBORN SCREENING ADVISORY COMMITTEE RECOMMENDATIONS

Background and Summary

Washington's newborn screening law (Chapter 70.83 RCW: Phenylketonuria and Other Preventable Heritable Disorders) declares that it is "…the policy of the state of Washington to make every effort to detect as early as feasible and to prevent where possible phenylketonuria and other preventable heritable disorders leading to developmental disabilities or physical defects." The statute delegates authority to the State Board of Health to determine which disorders in addition to phenylketonuria (PKU) are to be included in Newborn Screening required by the state. The statute also delegates authority to the Department of Health (DOH) to require screening tests of all newborn infants before they are discharged from the hospital for the detection of the disorders that are defined by the State Board of Health.

Current regulation (Chapter 246-650 WAC: Newborn Screening) requires screening for four disorders: PKU, congenital hypothyroidism, congenital adrenal hyperplasia, and hemoglobinopathies such as sickle cell disease. DOH conducts the testing for these disorders at the State Public Health Laboratory supported by a charge that is collected through the hospital or other birth facility.

Medical and technological advances in recent years have made it feasible to screen newborns for an increasing number of disorders. Many can be detected using the same dried blood specimen that is routinely collected to test infants for PKU and other disorders. The March of Dimes identifies ten core newborn screening tests that they advocate for including in every state newborn screening program. At the same time, Washington is the only state that does not screen its infants to detect galactosemia, a congenital metabolic problem that can cause sudden unexpected death or severe physical defects. Adding disorders to the panel currently required by Washington could further prevent illness and death through detection and treatment of affected newborns.

The State Board of Health, in conjunction with the State Department of Health, has been engaged in a deliberative process to consider amending the newborn screening rule (Chapter 246-650 WAC) to determine which, if any, additional disorders should be included in mandatory screening and to examine

the adequacy of existing privacy protections. The Board and the Department formed an advisory committee of experts to answer two questions: Is the current profile of screens adequate? If not, what principles or criteria do we need to apply to any additional screenings to be added? Board member Dr. Tom Locke and State Health Officer Dr. Maxine Hayes co-chaired the Newborn Screening Advisory Committee. Advisory committee members included representatives from a broad range of interested parties including parents, child health advocacy groups, medical specialists, hospitals, local public health officials, and public and private insurers. Examination of specific privacy protections was referred to the Board's Genetics Task Force that is completing a report to the Governor and Legislature by October 2002.

The Newborn Screening Advisory Committee has completed its charge and has prepared the following recommendations for the Board to consider.

Recommendations from Newborn Screening Advisory Committee

Criteria

The Newborn Screening Advisory Committee recommends that the Board adopt the proposed criteria to evaluate any additional mandated newborn screenings. The criteria fall into five categories: (1) prevention potential and medical rationale, (2) treatment available, (3) public health rationale, (4) available technology and (5) cost-benefit/cost-effectiveness.

Screenings

Based on the proposed criteria, the Newborn Screening Advisory Committee recommends that the Board consider adding the following six disorders to the screening requirements:

- Biotinidase Deficiency
- Galactosemia
- Homocystinuria
- Medium Chain Acyl Co-A Dehydrogenase Defincieny (MCADD)
- Maple Syrup Urine Disease (MSUD)
- Early Hearing Loss

The advisory committee considered adding cystic fibrosis to the recommended list of additional screenings. Although the committee does not recommend screening for cystic fibrosis to be added at this time, it strongly encourages the Board to re-evaluate this no later than spring 2004 or sooner if new data/science are available. Currently there are studies underway and a strong possibility exists that the new results will show improvements in therapeutic interventions and diagnostic technologies.

Options for Tandem Mass Spectrometry (MS/MS)

MS/MS technology is a new technology necessary to screen for MCADD, one of the disorders on the advisory committee's recommended list for mandatory screening. MS/MS can also be used for two other recommended disorders, homocystinuria and MSUD. However, MS/MS can detect a number of other metabolic disorders that may meet screening criteria but lack sufficient data at this time for adequate evaluation. Obtaining the additional information requires specific deliberate adjustment of the equipment. The Newborn Screening Advisory Committee considered three options if MS/MS were available:

Option 1: Limit screening to mandated disorders only.

Option 2: Make expanded screening an option available through DOH for those parents who want it (informed consent or dissent).

Option 3: Consider additional disorders by panel category including fatty acid oxidation, aminoacidemias, and organic acidemias detectable by MS/MS.

The advisory committee did not reach consensus on the options.

Option 1: 2 votes

Option 2: 0 votes

Option 3: 8 votes

Abstain: 3 votes

Reconvene Newborn Screening Advisory Committee

The Newborn Screening Advisory Committee recommends that the Board reconvene a group such as this advisory committee in one to two years to update the disorders and to make recommendations on the frequency of review of disorders. The advisory committee encourages the Board to maintain and utilize the DOH Newborn Screening report that is due to the Board on an annual basis.

Approved Board Action:

Dr. Locke and Dr. Hayes recommend that the Board:

(1) Accept the Newborn Screening Advisory Committee recommendations as part of the Board's consideration of amendments to Chapter 246-650 WAC.

(2) Adopt the Newborn Screening Advisory Committee's recommended timeline to complete the rule revisions by spring 2003.

(3) Request that the State Department of Health incorporate the Newborn Screening Advisory Committee's recommendations for the six additional disorders into its FY 2003-05 budget development process.

The following documents have been included to assist the Board in its discussion:

- List of Newborn Screening Advisory Committee Membership
- Chapter 70.83 RCW
- Chapter 246.650 WAC
- Proposed Statement of Inquiry for Chapter 246.650 WAC
- Matrix of Current Mandated Screenings
- Recommended Criteria
- Cost Benefit Analysis Summary of Screenings That Were Considered
- Final Matrix of Recommended Screenings
- Explanations of Disorders Recommended for Screening
- Timeline